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## SCID and Primary Immunodeficiency Fact Sheet

CIRM funds many projects seeking to better understand Severe Combined Immune Deficiency (SCID) and other primary immunodeficiency diseases to translate those discoveries into new therapies.

### Description

Primary immunodeficiencies are disease that compromise or destroy the immune system, leaving patients susceptible to serious infections. Typically, these diseases have genetic causes and many of them are rare. Severe combined immune deficiency or SCID is an example of a primary immunodeficiency.

SCID – also known as 'bubble boy disease' - is a rare genetic disorder, effecting one in 30,000 newborns. Left untreated the children die before the age of 2, and the only readily available treatment involves high-risk bone marrow transplants. Because these patients already have a compromised immune system, 10 to 20 percent don't survive the transplant.

Gene therapy has been used to correct the defect in certain types of SCID, but early gene modifying techniques resulted in some patients developing cancer. Newer gene therapy techniques appear to be safer but have been tried on fewer than 20 patients.

Researchers funded by California's stem cell agency are looking for a better alternative to help these children. They are trying to improve the safety of bone marrow transplant (BMT), which essentially uses the stem cells in bone marrow to give the children a new immune system that works properly. Most of the risk of current BMT procedures comes from the radiation or chemotherapy given to patients before the transplant to wipe out the patient's own stem cells that form immune cells. These regimens kill many types of cells beyond those intended and result in numerous toxic side effects.

### Clinical Stage Programs

#### Stanford School of Medicine (X-linked SCID)

This team proposes to replace SCID patients' dysfunctional immune cells with healthy ones using a safer form of bone marrow transplant (BMT). They plan to eliminate the bad cells with an antibody, a protein, that very specifically targets and eliminates blood forming stem cells. If successful, the procedure could open up similar BMT therapies to patients with other auto-immune diseases such as multiple sclerosis, lupus or diabetes that are generally not candidates for BMT currently. These diseases, while debilitating, are not immediately life-threatening and generally don't warrant the risks involved in BMT the way it is done today.

- [Read more about this project](#)
- [Read about this clinical trial](#)

#### St. Jude Children's Research Hospital

The team is using gene therapy to correct a genetic mutation in the blood stem cells of patients with X-linked SCID. The corrected stem cells are then transplanted back into the patient to restore their immune system's ability to produce healthy immune cells. This will allow the patient to fight off infections and will hopefully cure their disease.

- [Read more about this project](#)
- [Read about this clinical trial](#)

#### University of California, Los Angeles (Chronic granulomatous disease and ADA-SCID)

The team is developing a therapy for chronic granulomatous disease: a very rare primary immune deficiency disease that results in severe and recurrent infections that can impact quality and length of life. The UCLA team uses the patient's own genetically modified blood stem cells to create a new blood supply and immune system to eradicate the problem.

- [Read more about this project](#)
- [Read about this clinical trial](#)

UCLA is also developing a therapy for a form of SCID called adenosine deaminase (ADA)-deficient SCID. Patients with ADA-SCID lack an important enzyme called adenosine deaminase in their immune cells. Without this enzyme, toxic by-products build up in their immune cells and eventually kill them off leaving the patient susceptible to deadly infections. The group from UCLA will genetically modify patient blood stem cells to remove the disease-causing mutation and transplant these corrected stem cells back into the patient to create a new, healthy immune system.

- [Read more about this project](#)
- [Read about this clinical trial](#)

## CIRM Grants Targeting Immune Diseases including SCID

Researcher name	Institution	Grant Title	Grant Type	Approved funds	
Donald Kohn	University of California, Los Angeles	Efficacy and safety of cryopreserved autologous CD34+ HSC transduced with EFS lentiviral vector encoding for human ADA gene in ADA-SCID subjects	Clinical Trial Stage Projects	\$19,065,745	
Rosa Bacchetta	Stanford University	GENE EDITING FOR FOXP3 IN HUMAN HSC	Quest - Discovery Stage Research Projects	\$984,228	
Brian Sorrentino	St. Jude Children's Research Hospital	Lentiviral Gene Therapy for Infants with X-linked Severe Combined Immunodeficiency using Autologous Bone Marrow Stem Cells and Busulfan Conditioning	Clinical Trial Stage Projects	\$11,924,780	
Donald Kohn	University of California, Los Angeles	Primary Immune Deficiency Treatment Consortium (PIDTC) Annual Scientific Workshop	Conference II	\$29,807	
Jeanne Loring	Scripps Research Institute	Thymus based tolerance to stem cell therapies	Transplantation Immunology	\$1,108,921	

David DiGiusto	City of Hope	Development of RNA-based approaches to stem cell gene therapy for HIV	Early Translational II	\$3,097,160
Tippi MacKenzie	University of California, San Francisco	Maternal and Fetal Immune Responses to In Utero Hematopoietic Stem Cell Transplantation	Transplantation Immunology	\$1,230,869
John Zaia	City of Hope	ZINC FINGER NUCLEASE-BASED STEM CELL THERAPY FOR AIDS	Disease Team Research I	\$14,536,969
Anjana Rao	La Jolla Institute for Allergy and Immunology	Generation of regulatory T cells by reprogramming	Transplantation Immunology	\$1,464,446
Mark Walters	Children's Hospital of Oakland Research Institute	University of California, San Francisco (UCSF) CIRM Alpha Stem Cell Clinic	Alpha Stem Cell Clinics Network Expansion	\$7,999,999
Irvin Chen	University of California, Los Angeles	HPSC based therapy for HIV disease using RNAi to CCR5.	Disease Team Research I	\$9,905,604
Ellen Robey	University of California, Berkeley	Human Immune System Mouse models as preclinical platforms for stem cell derived grafts	Transplantation Immunology	\$1,005,605
Michele Calos	Stanford University	Safe, efficient creation of human induced pluripotent stem cells without the use of retroviruses	New Cell Lines	\$1,406,875
Judith Shizuru	Stanford University	Purified allogeneic hematopoietic stem cells as a platform for tolerance induction	Transplantation Immunology	\$1,233,275
Irvin Chen	University of California, Los Angeles	Genetic modification of the human genome to resist HIV-1 infection and/or disease progression	SEED Grant	\$616,800
Kenneth Weinberg	Stanford University	Engineered immune tolerance by Stem Cell-derived thymic regeneration	Transplantation Immunology	\$1,271,729
Thomas Lane	University of California, Irvine	Human Embryonic Stem Cells and Remyelination in a Viral Model of Demyelination	SEED Grant	\$368,081
Yang Xu	University of California, San Diego	Induction of immune tolerance to human embryonic stem cell-derived allografts	Transplantation Immunology	\$1,192,680
Kenneth Weinberg	Stanford University	Embryonic stem cell-derived thymic epithelial cells	SEED Grant	\$628,793
Judith Shizuru	Stanford University	A monoclonal antibody that depletes blood stem cells and enables chemotherapy free transplants	Disease Team Therapy Planning I	\$90,147
Morton Cowan	University of California, San Francisco	Gene Correction of Autologous Hematopoietic Stem Cells in Artemis Deficient SCID	Early Translational III	\$3,931,662

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Irving Weissman	Stanford University	Prospective isolation of hESC-derived hematopoietic and cardiomyocyte stem cells	Comprehensive Grant	\$2,471,386
Judith Shizuru	Stanford University	A monoclonal antibody that depletes blood stem cells and enables chemotherapy free transplants	Disease Team Therapy Development - Research	\$19,068,382
Samuel Pleasure	University of California, San Francisco	Human stem cell derived oligodendrocytes for treatment of stroke and MS	Comprehensive Grant	\$2,459,235
Mark Anderson	University of California, San Francisco	Generation of a functional thymus to induce immune tolerance to stem cell derivatives	Basic Biology V	\$1,191,000
Zack Jerome	University of California, Los Angeles	Human Embryonic Stem Cell Therapeutic Strategies to Target HIV Disease	Comprehensive Grant	\$2,401,903
John Adams	University of California, Los Angeles	UCLA-UCI Alpha Stem Cell Clinic (ASCC) Consortium	Alpha Stem Cell Clinics	\$8,000,000
Inder Verma	Salk Institute for Biological Studies	Curing Hematological Diseases	Early Translational I	\$5,979,252
Matthew Porteus	Stanford University	Pre-clinical development of gene correction therapy of hematopoietic stem cells for SCID-X1	Preclinical Development Awards	\$874,877
Mark Anderson	University of California, San Francisco	Stem cell differentiation to thymic epithelium for inducing tolerance to stem cells	Transplantation Immunology	\$1,314,089
Donald Kohn	University of California, Los Angeles	A Phase I/II, Non Randomized, Multicenter, Open-Label Study of G1XCGD (Lentiviral Vector Transduced CD34+ Cells) in Patients With X-Linked Chronic Granulomatous Disease	Clinical Trial Stage Projects	\$7,083,364
Jeffrey Bluestone	University of California, San Francisco	Stem cell tolerance through the use of engineered antigen-specific regulatory T cells	Transplantation Immunology	\$1,152,768
Gay Crooks	University of California, Los Angeles	Engineering Thymic Regeneration to Induce Tolerance	Transplantation Immunology	\$1,235,445
Jennifer Puck	University of California, San Francisco	Ex Vivo Transduction of the Human Artemis (DCLRE1C) cDNA by Lentiviral Vector AProArt into CD34+ Hematopoietic Cells for Artemis (ART)-Deficient Severe Combined Immunodeficiency (SCID)	Late Stage Preclinical Projects	\$4,268,865

Husein Hadeiba	Palo Alto Institute for Research and Education	Application of Tolerogenic Dendritic Cells for Hematopoietic Stem Cell Transplantation	Transplantation Immunology	\$733,061	
					Total: \$141,327,802.00

## Resources

- The Stem Cellar blogs about SCID and immune disease research
- Immune Deficiency Foundation
- NIH: SCID Fact Sheet
- SCID.net
- Find a clinical trial near you: NIH Clinical Trials database
- Family Caregiver Alliance
- National Family Caregivers Association

### Find Out More:

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